GENOMENON

For Immediate Release

Genomenon Accelerates Gene and Variant Curation With Launch Of Mastermind

New Product Dramatically Accelerates Genetic Variant Curation – Speeding Time to Diagnosis

Ann Arbor, Mich - February 13, 2017 - Genomenon announces the launch of Mastermind, a novel analytic and data visualization tool that accelerates gene and variant curation by providing immediate insight into millions of prioritized scientific articles from the primary medical literature. Mastermind delivers a comprehensive knowledge base of disease-gene-variant relationships curated from primary medical literature, organized into clinical categories, and prioritized by the strength of evidence through our automated platform and algorithms to improve genetic diagnosis for patients with cancer and other genetic diseases.

The challenge faced by clinicians when conducting genetic variant interpretation is that only a small fraction of the disease-gene-variant relationships found in the full-text of the scientific publications are captured in the titles or abstracts that PubMed searches. Mastermind solves this problem by finding and prioritizing the primary literature for any given disease-gene-variant combination through its full-text search of many millions of scientific articles. This reduces the time pathologists and geneticists spend searching for articles for variant curation by up to 80% and improves the accuracy of these interpretation.

Genomenon founder, Mark Kiel MD, PhD, experienced first-hand the time it takes geneticists and pathologists to search through medical publications for disease, gene and variant relationships and developed Mastermind to address the problem. “As a molecular pathologist at the University of Michigan, 80% of my day was spent searching for information on PubMed, Google, COSMIC and HGMD and only 20% of my time was spent on interpreting the data” said Dr. Mark Kiel. “We developed Mastermind to allow highly trained clinicians to spend less time on inefficient and incomplete literature searches and let them focus their skills on interpreting the data for patient reports.”

In developing Mastermind, Genomenon has spent the last three years poring through 3.3 million full-text articles and found every disease-gene-variant combination discussed in the literature covering somatic cancer, hereditary cancer, cardiomyopathy, and infertility. Mastermind has reviewed literature for all the genes from ThermoFisher’s Ion AmpliSeq comprehensive cancer panel, Illumina’s TruSight Tumor 170 solid tumor cancer panel, ThermoFisher’s and Fulgent’s hereditary cancer panels, and ThermoFisher’s, Illumina’s and Fulgent’s cardiomyopathy panels. Genomenon has found all the variants cited in the literature associated with each of these disease-gene combinations. The company is continuously expanding its database and plans to add over 3 million more articles covering additional genetic diseases over the next 6 months to assemble the most comprehensive database of genomic knowledge ever created.

About Genomenon:
Genomenon has eliminated the manual search process for gene and variant curation with a scientific literature database and visualization software of prioritized evidence for use in clinical decision-making. We have mined the full text of millions of genomic-related scientific articles to create a comprehensive genomic knowledge

base that enables pathologists and geneticists to quickly and accurately curate disease-causing variants from genomic-sequencing datasets. With the comprehensive data set and deep insight into all of the genomic literature, we're able to provide fascinating results – from accelerated gene and variant curation to evidence-based panel design – by scouring the literature to find genes and variants linked to specific diseases within the literature. For more information, visit www.genomenon.com or email sales@genomenon.com.

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